Biotinidase Deficiency

Biotinidase deficiency is an autosomal recessive disorder that interferes with the body's ability to synthesize biotin, a B vitamin. Biotinidase is an enzyme responsible for recycling biotin in the degradation of carboxylases, as well as freeing the protein bound form in digestion. Biotin is an essential co-factor in several metabolic pathways, the deficiency of which ultimately results in neurologic damage.

Estimated Incidence (MI):	1: 34,000 (includes profound and partial deficiencies)
Laboratory Screening Test:	Colorimetric assay used to detect enzyme activity
Timing of Test:	Valid at birth
Feeding Effect:	None
Transfusion Effect:	Transfusion of whole blood may interfere with the accuracy of testing, causing a false negative result. **Obtain newborn screen before transfusion.**
Confirmation:	All strong and persistent borderline positive tests are referred to the Pediatric Neurology metabolic Clinic (PNMC) for confirmation (734) 763-4697. **Do not send diagnostic labs before contacting the PNMC.**
Treatment:	Daily oral biotin supplement

For further information, contact the Newborn Screening Program
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